BIRTH DEFECT RISK FACTOR SERIES: TRISOMY 13 (PATAU SYNDROME)

DEFINITION

Trisomy 13 (Patau syndrome) is the third most common autosomal abnormality among live births after Down syndrome (trisomy 21) and Edwards syndrome (trisomy 18). Most trisomy 13 cases result from total trisomy 13 (Buyse, 1990). A small proportion of trisomy 13 cases result from mosaicism and translocation (Forrester and Merz, 1999; Carothers et al., 1999; Buyse, 1990). Some trisomy 13 fetuses detected in mid-trimester do not survive to term (Hook et al., 1989).

Maternal serum screening has not been found to differ significantly between pregnancies affected with trisomy 13 and normal pregnancies (Saller and Canick, 1999; Canick and Saller, 1993). Over the last several decades, prenatal ultrasonography can detect a variety of structural anomalies frequently associated with trisomy 13 (Abramsky and Chapple, 1993; Vintzileos et al., 1987). Prenatal ultrasonography and definitive diagnosis by karyotyping through such procedures as amniocentesis and chorionic villus sampling have allowed trisomy 13 to be identified in utero. Studies from various birth defects surveillance systems have found that, in regions where elective termination is allowed, prenatal diagnosis and elective termination reduce the prevalence of trisomy 13 (Chaabouni et al., 2001; De Vigan et al., 2001; Forrester and Merz, 1999; Carothers et al., 1999; Forrester et al., 1998; Riley et al., 1998; Wyllie et al., 1994; Abramsky and Chapple, 1993; Pradat et al., 1991).

ETIOLOGY

Trisomy 13 involving total trisomy 13 results from nondisjunction, usually in formation of the eggs or sperm, where one of the gametes ends up with an extra chromosome 13. Nondisjunction may occur in the first meiotic stage (MI) or the second meiotic stage (MII). The extra chromosome 13 is of maternal origin in 88 percent of the cases and of paternal origin in 12 percent of the cases. Among trisomy 13 cases of maternal origin, almost all result from nondisjunction in MI (Nicolaidis and Petersen, 1998; Zaragoza et al., 1994).

DEMOGRAPHIC AND REPRODUCTIVE FACTORS

Risk of trisomy 13 is well known to increase with increasing **maternal age** (Forrester and Merz, 1999; Carothers et al., 1999; Goldstein and Nielsen, 1988; Schreinemachers et al., 1982). Trisomy 13 risk has been associated with increasing **paternal age** (Baty et al., 1994); however, once maternal age is taken into consideration the association with paternal age tends to disappear. Higher trisomy 13 risk has been associated with increasing **parity** (Monteleone et al., 1981); however, this observation may be confounded by maternal age.

Race/ethnicity has not been reported to influence trisomy 13 rates (Buyse, 1990). One study found that, of the four racial/ethnic groups examined (white, Far East Asian, Pacific Islander, Filipino), trisomy 13 risk was highest for Far East Asians and lowest for Pacific Islanders (Forrester and Merz, 1999). However, the differences in risk appeared to be due to differences in maternal age distribution among the racial/ethnic groups. Another investigation observed no significant difference in risk of Patau syndrome in infants born to Vietnamese women compared with infants born to non-Hispanic white women in California (Shaw et al., 2002).

Geographic area may influence trisomy 13 risk. One study reported higher trisomy 13 rates among urban

residents (Forrester and Merz, 1999). However, this high risk among urban residents seemed to be due to differences in maternal age distribution between urban and rural areas.

One investigation has reported a **secular trend** for trisomy 13, with the prevalence of the aneuploidy increasing over time. However, the increase in trisomy 13 prevalence over time was considered due to increasing numbers of births to older women and increasing prenatal diagnosis of affected pregnancies (Forrester and Merz, 1999). There does not appear to be **seasonal variation** in trisomy 13 rates (Videbech and Nielsen, 1984).

Infant sex influences the risk for trisomy 13. Males are more likely than females to have the aneuploidy (Forrester and Merz, 1999; Carothers et al., 1999; Riley et al., 1998; Goldstein and Nielsen, 1988; Monteleone et al., 1981). Trisomy 13 is also associated with lower **birth weight**, **prematurity**, and **intrauterine growth retardation** but not **plurality** (Rasmussen et al., 2001; Riley et al., 1998; Mili et al., 1991).

The **recurrence risk** for trisomy 13 has been reported to be approximately 1% (Baty et al., 1994).

FACTORS IN LIFESTYLE OR ENVIRONMENT

No lifestyle or environmental factors have been definitively reported to affect trisomy 13 risk.

One study has reported that women who had infants or fetuses with trisomy 13 were not more likely to have mutation in the **methylenetetrahydrofolate reductase (MTHFR) gene** or the **methionine synthase reductase (MTRR) gene** (Hassold et al., 2001).

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Please Note: *The primary purpose of this report is to provide background necessary for conducting cluster investigations. It summarizes literature about risk factors associated with this defect. The strengths and limitations of each reference were not critically examined prior to inclusion in this report. Consumers and professionals using this information are advised to consult the references given for more in-depth information.*

This report is for information purposes only and is not intended to diagnose, cure, mitigate, treat, or prevent disease or other conditions and is not intended to provide a determination or assessment of the state of health. Individuals affected by this condition should consult their physician and when appropriate, seek genetic counseling.